

Eight years of growth hormone treatment in a patient with Schaaf-Yang Syndrome

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Conclusion

Eight years of GH-treatment improved height-SDS, BMI-SDS and fat mass percentage in a patient with SYS

Background

Schaaf-Yang syndrome (SYS) is a rare disorder caused by a truncating mutation in the gene *MAGEL2*, located in the Prader-Willi syndrome (PWS) critical region on chromosome 15. SYS is characterized by neonatal hypotonia, feeding problems in early infancy and intellectual disability, obesity and behavioral problems throughout childhood, resembling a PWS phenotype. In this case report we describe a 15-year-old girl, receiving growth hormone (GH) treatment since the age of 6 years, because of partial GH deficiency. She was diagnosed with SYS at 12 years of age.

Methods

Medical records were retrospectively reviewed. Data on height, weight and BMI were available for the entire eight years of GH treatment. Data on body composition were only available for the most recent four years since she was diagnosed with SYS.

Results

The patient was extensively evaluated by the Dept. of Clinical Genetics and Pediatric Neurology, because of psychomotor delay. Whole exome sequencing (WES) eventually showed a frameshift, de novo mutation in the *MAGEL2* gene. After 8 years of GH treatment, height SDS had increased from -3.95 to -1.11. BMI improved from +1.53 SDS to -0.09 SDS. After being diagnosed with SYS, she received multidisciplinary care by the Dutch PWS team. In these 4 years, fat mass percentage (fat%) decreased from 44% to 31.2% (a decrease from 1.78 SDS to 0.57 SDS) and lean body mass (LBM) increased from -2.96 SDS to -2.06 SDS.

BMI for age

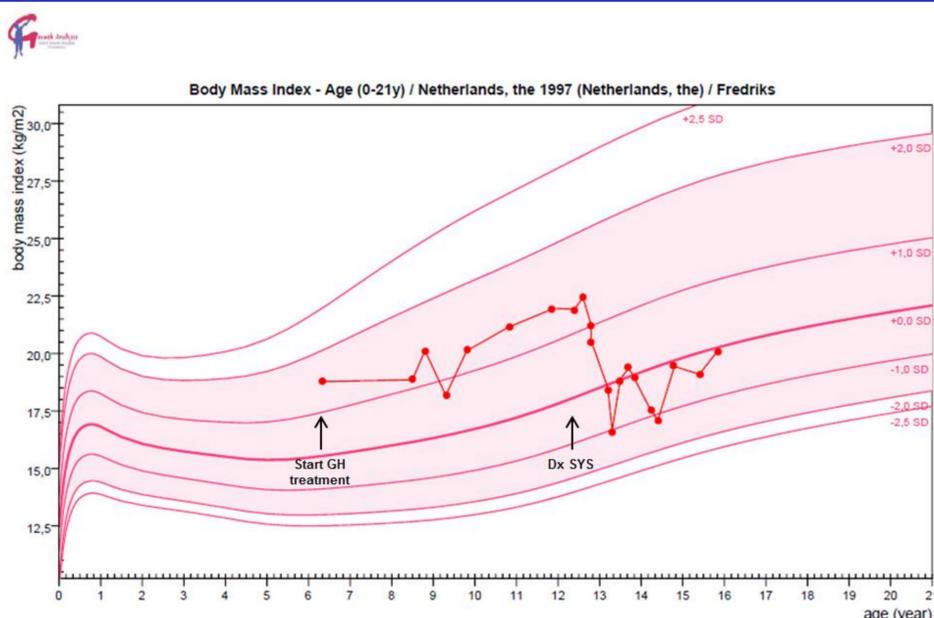


Fig. 1: BMI for age shows a decrease in BMI from +1.52 SDS at onset to -0.09 in the most recent 4 years of GH treatment and multidisciplinary care.

Height for age

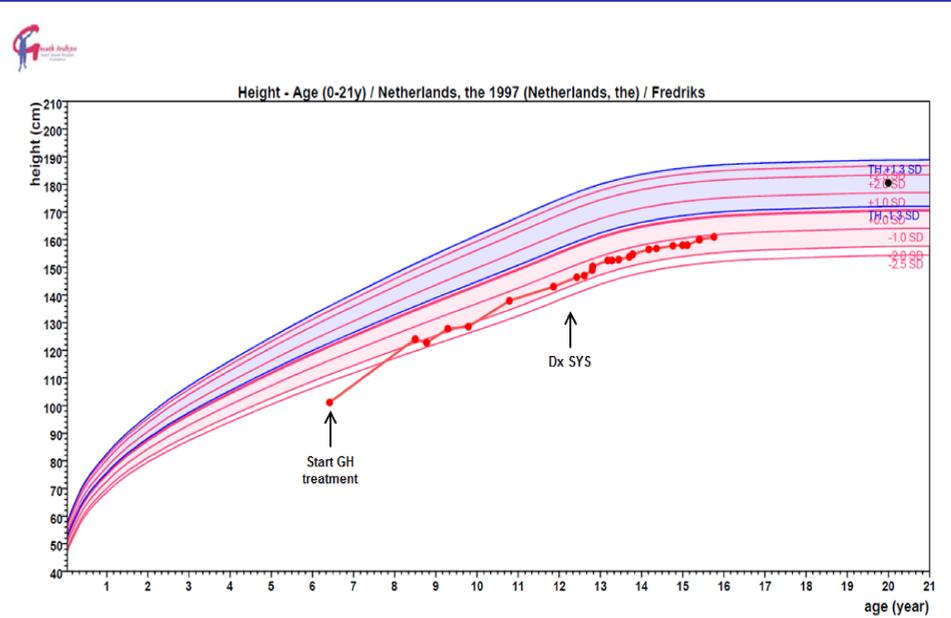


Fig. 2: Height for age shows an increase in height from -3.95 SDS to -1.11 SDS during 8 years of GH treatment.

Discussion

This patient with SYS shows a normalization of height SDS, a significant decrease in fat% SDS and increase of LBM SDS during 8 years of GH treatment without any side effects, suggesting that GH might be efficacious and safe for patients with SYS. Further studies need to confirm the effectiveness of GH treatment in other patients with SYS.

